Flecked-retina Disorders: Deletion/Duplication Panel

**Test Code:** MD134  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81479 x1

### Condition Description

Flecked-retina refers to a varying distribution and size of yellowish lesions within the retina (optic nerve or vascular anomalies are not generally observed). Flecked-retina disorders may be stationary or progressive and range from benign to visually debilitating. The term "flecked-retina" is generally not favored by retina specialists due to the distinctive pattern and clinical course of each individual disorder. Examples of conditions in this category include benign flecked-retina, fundus flavimaculatus (Stargardt's), and fundus albipunctatus.

### References:
- OMIM  
- GeneReviews  
- Emory and Rimoin's Principles and Practice of Medical Genetics, 5th Edition

### Genes

**ABCA4, PLA2G5, PRPH2, RDH5, RHO, RLBP1**

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of flecked-retina disorders.  
- Carrier testing in adults with a family history of flecked-retina disorders.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### Specimen Requirements

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:
- In EDTA (purple top) tube: Infants (2 years): 3-5 ml  
  Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:
- In microtainer: 10 ug

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Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Special Instructions**

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical correlation with test results.

**Related Tests**

- Eye Disorders: Comprehensive Sequencing and Deletion/Duplication Panels.
- Flecked-retina Disorders: Sequencing Panel.